SLW 12/7/04

Human Chromosome 16 Phosphatidylinositol-like Polypeptide

~~HUMAN CHROMOSOME 15 AND 16 BARDET-BIEDL SYNDROME POLYNUCLEOTIDES AND POLYPEPTIDES AND METHODS OF USE~~

TECHNICAL FIELD

This invention relates to the identification and recombinant expression of human chromosome 15 and 16 Bardet-Biedl Syndrome Region (BBSR) proteins.

BACKGROUND OF THE INVENTION

Bardet-Biedl Syndrome (BBS) is a clinically and genetically heterogeneous autosomal recessive disorder characterized by obesity, polydactyly, hypogenitalism, retinal degeneration, mental retardation and heart and kidney abnormalities.

Elbedour et al. (1994) *Am. J. Med. Genet. 52(2)*:164-169 have reported hypertrophy of the interventricular septum and dilated cardiomyopathy, in addition to other previously reported congenital heart defects associated with BBS.

BBS has been mapped to loci on several human chromosomes including chromosomes 3, 11, 15 and 16. These loci include the 3p12, 11q13, 15q22 and the 16q21 chromosomal sites; also referred to as the BBS3, BBS1, BBS4 and BBS2 loci, respectively. (Bruford et al. (1997) *Genomics 41(1)*:93-9; Leppert et al. (1994) *Nature Genet. 7*:108-112; Carmi et al. (1995) *Hum. Mol. Genet. 4*:9-13; Kwitek-Black et al. (1993) *Nature Genet. 5*:392-396; Sheffield et al. (1994) *Hum. Mol. Genet. 3*:1331-1335; Beales et al. (1997) *J. Med. Genet. 34(2)*:92-8.).

Attempts to associate particular phenotypes with particular BBS loci have been reported. For example, Beales et al. (1997) *J. Med. Genet. 34(2)*:92-8, reported that affected subjects linked to the BBS2 and 4 loci were significantly shorter than their parents, while those linked to the BBS1 locus were taller, indicating possible role for various BBS genes in influencing growth characteristics such as height. Carmi et al. (1995) *Hum. Mol. Genet. 4*:9-13, reported that BBS3 is associated with polydactyly of all four limbs while